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14. ABSTRACT

As the aviation population ages, it increasingly becomes affected by neurological diseases that may cause disability and reduce mobility and freedom, both mentally and physically. While some disorders progress slowly with relatively little effect, others may result in significant motor and neurological deficits that impair the ability to effectively perform aviation tasks. New advances in disease testing and diagnosis, such as genetic testing, now provide increased means for disease diagnosis but also possible therapeutic treatments. Indeed, according to some experts, genetic testing and therapy may be key to future disease detection, therapy, and even prevention. In this case report, a second-class airman with the gene that causes Huntington's disease will be discussed, as well as the aeromedical concerns associated with its long-term management.

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Huntington's Disease

Case Report, by Robert Craig-Gray, MD

Summary

As the aviation population ages, it increasingly becomes affected by neurological diseases that may cause disability and reduce mobility and freedom, both mentally and physically. While some disorders progress slowly with relatively little effect, others may result in significant motor and neurological deficits that impair the ability to effectively perform aviation tasks. New advances in disease testing and diagnosis, such as genetic testing, now provide increased means for disease diagnosis but also possible therapeutic treatments. Indeed, according to some experts, genetic testing and therapy may be key to future disease detection, therapy, and even prevention. In this case report, a second-class airman with the gene that causes Huntington's disease will be discussed, as well as the aeromedical concerns associated with its long-term management.

History

A 40-YEAR-OLD COMMERCIAL pilot with 4,000-plus flying hours presented to the office of his aviation medical examiner (AME) for re-issuance of his second-class medical certificate. As a former USAF pilot, the airman held a third-class medical certificate and student medical certificate obtained during his primary pilot training. After separating from the service approximately 2 years ago, the airman underwent an initial employment interview and medical screening exams for a regional commuter airline, which he passed, and he was subsequently hired as a co-pilot. He has been flying for 16 months without incident and reported no problems with his recent change in career and lifestyle. The airman did not smoke and reported moderate alcohol consumption with no history of abuse or dependence and reported no significant medical or surgical history, other than elevated cholesterol, which he controlled with diet and physical activity. He had no recent hospitalizations or significant illnesses. The airman's maternal grandfather died in his early 50s from an unknown disease, but both parents are living and healthy. He has 2 siblings (1 sister and 1 brother) who are both younger and otherwise healthy and disease-free.

During further AME review of his FAA Form 8500-8, it was noted that the airman had checked positive for block 18x and annotated Huntington's gene in the explanation block. Upon further questioning, he stated that his 30-year-old sister recently underwent genetic testing associated with her first pregnancy, during which she was offered advanced genetic screening for common familial diseases. She was found to be positive for the genetic marker associated with Huntington's disease. After receiving further genetic counseling, she shared this information with the airman and the rest of her family, all of whom agreed to undergo confirmatory testing due to the inheritance pattern of the disease. The airman's father and brother were both negative for the Huntington gene; however, both the airman and his mother were found to be positive and underwent further genetic counseling. The airman had discussed the results and his concern with his personal physician but otherwise reported no disability or medical concerns. His AME requested deferred approval of his second-class certification to the FAA regional flight surgeon.

Aeromedical Concerns

The presence of neurological disease in an airman presents a unique challenge to the aviation medical examiner. He/she must weigh the degree of motor or sensory deficits (if present) and the prognosis versus the need for safety in the likelihood of sudden incapacitation or progressive decline, as well as consideration for the airman's right to fly. According to the *Guide for Aviation Medical Examiners*, "A history or the presence of any neurological condition or disease that potentially may incapacitate an individual should be regarded as initially disqualifying. Issuance of a medical certificate to an applicant in such cases should be denied or deferred, pending further evaluation. Applications from individuals with potentially disqualifying conditions should be forwarded to the FAA Aerospace Medical Certification Division."[1]

In cases of neurological disease, additional information is helpful and should include all additional medical records and history, any specialty consultation reports, along with appropriate laboratory and radiological imaging studies. Reports should detail the history of any symptoms or disturbances due to the airman's underlying condition. Special attention should be given to anything that may be acutely incapacitating in an aviation environment such as pain, weakness, vertigo or incoordination, seizures or a disturbance of consciousness, visual disturbance, or mental confusion. Additional attention should be given to any prognosis that may be chronic and progressive and/or incompatible with safe aircraft operation either due to the severity of neurologic deficit, its psychological impairment, or future unpredictability.

In this case, with respect to Huntington's disease, key points critical to airman certification include: 1) the presence of active symptoms of neurological disease, 2) the presence of mental health disorders such as depression or dementia, and 3) the ability to satisfactorily monitor the airman for progression of the disease. While medications and cognitive/psychological and physical therapy can help reduce symptoms and slow disease progression, individuals testing positive for the disease and who develop symptoms face a 100% mortality rate and endure progressive, debilitating disease. Although only 50% of those testing positive for the disease actually develop symptoms, currently there is no further predictive capacity to determine who will become ill and who will remain disease-free, and this must also be accounted for in any aeromedical decision-making. Therefore, an FAA medical certificate should not be issued to any applicant who tests positive for Huntington's disease under such criteria without first consulting with an FAA Regional Flight Surgeon or the FAA Aerospace Medical Certification Division (AMCD).

Outcome

A medical status report was obtained from the airman's neurologist and revealed no detectable neurological disease or disability. Additional requested neuropsychiatric testing

Continued

Etiology of Huntington's disease

Huntington's disease is a rare, progressive, degenerative disease that results in severe neurologic disease, disability, and eventually death in affected individuals. Until recently, the diagnosis of Huntington's disease was based on physical symptoms and a family history of the disease, but modern genetic testing can now detect the defect in the HTT allele of chromosome #4.[3] In affected individuals, errors in DNA replication occur that result in multiple repeats of CAG expression sequences on the affected chromosome, which may also be passed down to offspring.[3,4] It is an autosomal dominant disorder where only one copy of the defective gene inherited from either parent is necessary to produce disease. If one parent possesses the single defective gene, the chance that an offspring will have the defect is 50%. Worldwide, the prevalence of the disease is 5-10 cases per 100,000 persons but varies geographically.[3] Prevalence is similar for both men and women.[3]

Early onset of disease represents a small number of cases and adult onset Huntington's disease represents the majority of cases seen. Symptoms usually become evident during the mid-30s to 40s but may be seen earlier in individuals affected with larger numbers of repeating CAG expression sequences.[4,5,6] In these individuals, more severe and rapid disease progression occurs. Often, family and friends notice changes associated with Huntington's disease prior to those affected with the disease.[5,6] These symptoms can include antisocial behavior, hallucinations, paranoia, psychosis, and personality changes.[6] These early symptoms may be seen prior to the development of motor symptoms, which occur later in the progression of the disease. Motor symptoms resemble those seen in Parkinson's disease and include rigidity, unsteady gait, quick uncontrolled movements, clumsiness, and tremors.[6] Brain MRI or PET scans often show changes associated with neurological damage as nerve cells within the brain waste away, die, or degenerate.[6]

The goal of treatment in Huntington's disease is currently focused on slowing its insidious progression and minimizing disability. Increasingly, respiratory difficulties, speech and motor impairment, depression, and difficulty swallowing are complications seen in the final stages of the disease, which limit the daily activities of these patients.[6] Medication therapy may vary depending on disease symptoms and includes dopamine blockers, such as tetrabenazine, to help reduce abnormal movements and haloperidol, to minimize violent outbursts and hallucinations, or amantadine to control extra movements.[6,7] Some evidence also suggests that co-enzyme Q10 or gene therapy may help slow disease progression.[8] Side effects from many drugs used to treat the Huntington's disease include hyper-excitability, fatigue, and restlessness. Mental disorders commonly seen among persons affected with Huntington's disease include depression and suicide and should be monitored for and treated.[3,4,5] As the disease progresses, patients require increased assistance and may eventually require 24-hour care.[5,6] Unfortunately, no cure currently exists for the disease, and death usually occurs 15 to 20 years after the initial diagnosis. Infections and suicide are the most common causes of death in individuals with the disease.

showed no changes in his cognitive abilities or evidence of psychiatric disease. Laboratory and radiographic testing were also normal. Under 14 CFR part 67.213 (b)(c), 67.313 (b)(c), and 67.209 (b), and 67.309 (b), an authorization for special issuance was subsequently granted for this airman. Given the unexpected nature and terminal prognosis of Huntington's disease, the airman should also report any changes in his medical condition immediately to the FAA and cease aviation operations per Title 14 CFR §61.53, which restricts operation of an aircraft with any medical [known] deficiency.[2]

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About the author. *Robert Craig-Gray, MD, MPH, LtCol, USAF, MC, SFS, was a Resident in Aerospace Medicine when he wrote this case report at the Civil Aerospace Medical Institute. Currently, he is a Resident in Occupational Medicine with the U.S. Air Force School of Aerospace Medicine at Wright Patterson AFB, Ohio.*